SYNE1 gene

spectrin repeat containing nuclear envelope protein 1

Normal Function

The *SYNE1* gene provides instructions for making a protein called Syne-1 that is found in many tissues, but it seems to be especially critical in the brain. The Syne-1 protein plays a role in the maintenance of the part of the brain that coordinates movement (the cerebellum). The Syne-1 protein is active (expressed) in Purkinje cells, which are located in the cerebellum and are involved in chemical signaling between nerve cells (neurons). The protein is thought to attach the membrane of Purkinje cells to the actin cytoskeleton, which is a network of fibers that make up the cell's structural framework. It is not clear what role this attachment plays in Purkinje cell function.

Health Conditions Related to Genetic Changes

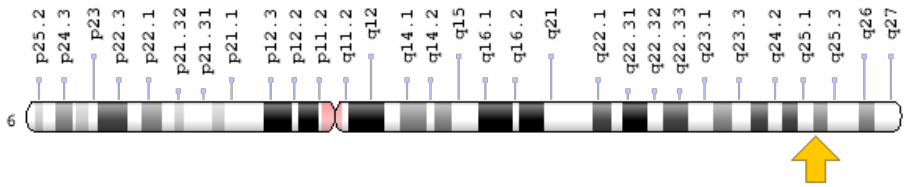
autosomal recessive cerebellar ataxia type 1

At least seven mutations in the *SYNE1* gene have been found to cause autosomal recessive cerebellar ataxia type 1 (ARCA1). All the mutations that have been identified create a premature stop signal in the instructions for making the Syne-1 protein, resulting in an abnormally short protein with impaired function. A dysfunctional Syne-1 protein is thought to impair Purkinje cell function and disrupt signaling between neurons in the cerebellum. The loss of brain cells in the cerebellum causes the movement problems characteristic of ARCA1, but it is unclear how this cell loss is related to impaired Purkinje cell function.

Chromosomal Location

Cytogenetic Location: 6q25.2, which is the long (q) arm of chromosome 6 at position 25.2

Molecular Location: base pairs 152,121,684 to 152,637,399 on chromosome 6 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- ARCA1
- MYNE1
- myocyte nuclear envelope protein 1
- Nesp1
- nesprin-1
- nuclear envelope spectrin repeat protein 1
- spectrin repeat containing, nuclear envelope 1
- SYNE1_HUMAN

Additional Information & Resources

Educational Resources

- Neuroscience (second edition, 2001): Cerebellar Circuitry and the Coordination of Ongoing Movement
  https://www.ncbi.nlm.nih.gov/books/NBK10840/
- Washington University, St. Louis: Neuromuscular Disease Center
  http://neuromuscular.wustl.edu/ataxia/recatax.html#ataxsyne1

GeneReviews

- SYNE1-Related Autosomal Recessive Cerebellar Ataxia
  https://www.ncbi.nlm.nih.gov/books/NBK1379

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28SYNE1%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+2880+days%22%5Bdp%5D

OMIM

- SPECTRIN REPEAT-CONTAINING NUCLEAR ENVELOPE PROTEIN 1
  http://omim.org/entry/608441

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_SYNE1.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=SYNE1%5Bgene%5D
- HGNC Gene Family: Spectrin repeat containing nuclear envelope family
  http://www.genenames.org/cgi-bin/genefamilies/set/1252
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=17089
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/23345
- UniProt
  http://www.uniprot.org/uniprot/Q8NF91

Sources for This Summary

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  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17503513
- GeneReview: SYNE1-Related Autosomal Recessive Cerebellar Ataxia
  https://www.ncbi.nlm.nih.gov/books/NBK1379
- Gros-Louis F, Dupré N, Dion P, Fox MA, Laurent S, Verreault S, Sanes JR, Bouchard JP, Rouleau GA. Mutations in SYNE1 lead to a newly discovered form of autosomal recessive cerebellar ataxia. Nat Genet. 2007 Jan;39(1):80-5. Epub 2006 Dec 10.
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